GENETIC REFERENCE MATERIALS

Abstract of the Disclosure

The invention provides a genetic reference standard with at least one human genetic reference sequence (having a human DNA sequence containing at least one genetic variant whose presence in the DNA of a human subject is indicative of a pathological condition, a predisposition to a pathological condition, or a predisposition to an adverse reaction to external stimuli, or is indicative of a patient's likely response to a therapeutic intervention, i.e. a variant used in pharmacogenomic analysis) cloned into a non-mammalian animal cell line. There are also provided such reference standards where the human DNA is targeted to specific location in the host genome, using homologous recombination. The invention further provides a method of detecting a genetic variant using such reference standards.